SLC37A4 gene

solute carrier family 37 member 4

Normal Function

The *SLC37A4* gene provides instructions for making a protein called glucose 6-phosphate translocase. This protein transports the sugar molecule glucose 6-phosphate from the fluid inside the cell (cytoplasm) to the endoplasmic reticulum, which is a structure inside cells that is involved in protein processing and transport. At the membrane of the endoplasmic reticulum, glucose 6-phosphate translocase works together with the glucose 6-phosphatase protein (produced from the *G6PC* gene) to break down glucose 6-phosphate. The breakdown of this molecule produces the simple sugar glucose, which is the primary energy source for most cells in the body.

Health Conditions Related to Genetic Changes

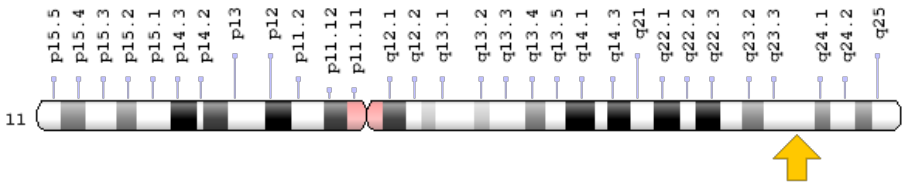
glycogen storage disease type I

More than 80 mutations in the *SLC37A4* gene have been found to cause glycogen storage disease type Ib (GSDIb). Most of these mutations change single protein building blocks (amino acids) in glucose 6-phosphate translocase. *SLC37A4* gene mutations disrupt the normal functioning of glucose 6-phosphate translocase and prevent the transport of glucose 6-phosphate to the endoplasmic reticulum. If glucose 6-phosphate cannot get to the endoplasmic reticulum, it cannot get broken down and glucose is not produced. Glucose 6-phosphate that is not broken down to glucose is converted to fat and glycogen, a complex sugar that is stored within cells. Too much fat and glycogen stored within a cell can be toxic. This buildup damages organs and tissues throughout the body, particularly the liver and kidneys, leading to the signs and symptoms of GSDIb. For reasons that are unclear, mutations in the *SLC37A4* gene also cause a shortage of white blood cells (neutropenia) in people with GSDIb.

Chromosomal Location

Cytogenetic Location: 11q23.3, which is the long (q) arm of chromosome 11 at position 23.3

Molecular Location: base pairs 119,024,351 to 119,030,906 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- G6PT1
- G6PT1_HUMAN
- glucose-6-phosphate translocase
- solute carrier family 37 (glucose-6-phosphate transporter), member 4

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Generation of Glucose from Glucose 6-Phosphate (image)
  https://www.ncbi.nlm.nih.gov/books/NBK22591/figure/A2279/

GeneReviews

- Glycogen Storage Disease Type I
  https://www.ncbi.nlm.nih.gov/books/NBK1312

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SLC37A4%5BTIAB%5D%29+OR+%28glucose-6-phosphate+translocase%5BTIAB%5D%29%29+OR+%28G6PT1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- SOLUTE CARRIER FAMILY 37 (GLUCOSE-6-PHOSPHATE TRANSPORTER), MEMBER 4
  http://omim.org/entry/602671

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SLC37A4.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC37A4%5Bgene%5D
- HGNC Gene Family: Solute carriers
  http://www.genenames.org/cgi-bin/genefamilies/set/752
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4061
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/2542
- UniProt
  http://www.uniprot.org/uniprot/O43826

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